

Simeon Antonov Boyadjiev Boyd, M.D.

Philosophy of Care

Birth defects are the leading cause of infant mortality in the United States, accounting for more than 20% of all infant deaths, yet the causes of about 70% of all birth defects are still unknown. Dr. Boyd has dedicated his career to the care of children affected with congenital genetic anomalies and to providing genetic counseling and support to their families. Using a "bed-to-bench-and back" approach he has characterized several previously unknown genetic syndromes, identified the causative genes, and used this knowledge for optimal clinical care of the affected patients.

Clinical Interests

Dr. Boyd is a clinical geneticist and pediatrician involved in studies of birth defects, such as nonsyndromic craniosynostosis, bladder epispadias-exstrophy complex, and cleft lip and/or palate. He leads the International Craniosynostosis Consortium (<https://health.ucdavis.edu/pediatrics/research/labs/boyd-genetics-lab/>) and has been continuously funded by NIH NIDCR grants since 2000. His group has recruited more than 1,500 families with these birth defects for clinical and molecular studies.

Research/Academic Interests

Dr. Boyd's clinical interests are focused on diagnostics and longitudinal clinical characterization of congenital birth defects and syndromes with multiple congenital anomalies. Dr. Boyd also leads a molecular genetics research laboratory (<https://health.ucdavis.edu/pediatrics/research/labs/boyd-genetics-lab/>) that is instrumental for the identification of the genes causing nonsyndromic craniosynostosis and several related genetic conditions. Using Genome-wide association studies (GWAS) and Whole-genome sequencing (WGS) Dr. Boyd has contributed to the identification of genes and environmental factors contributing to the risk of craniosynostosis and other birth defects.

Title Professor, Department of Pediatrics

Specialty Clinical Genetics

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Division General Pediatrics

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Languages Bulgarian, Russian



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Education	M.D., Pediatrics, Leningrad Pediatric Medical Institute, Leningrad, Russia 1985
Residency	Pediatrics, State University of New York (SUNY), New York NY 1995-1997
Fellowships	Clinical Genetics, The Johns Hopkins University, Baltimore MD 1997-2000
Board Certifications	American Board of Medical Genetics - Clinical Genetics (M.D.) American Board of Pediatrics
Professional Memberships	American College of Medical Genetics and Genomics European Society of Human Genetics
Honors and Awards	Dean Nomination Clinical Research Forum Award, UC Davis, 2012 Dean nomination for Provost Fellow, UC Davis, 2007 Keynote Speaker, COAST meeting "Developmental defects of the Craniofacial Skeleton", 2007 President Elect of the Society of Craniofacial Genetics, 2002, 2003, 2004 Vice President Elect of the Society of Craniofacial Genetics, 2000, 2001, 2002
Select Recent Publications	Justice CM, Musolf AM, Cuellar A, Lattanzi W, Simeonov E, Kaneva R, Paschall J, Cunningham M, Wilkie AOM, Wilson AF, Romitti PA, Boyadjiev SA. Targeted Sequencing of Candidate Regions Associated with Sagittal and Metopic Nonsyndromic Craniosynostosis. <i>Genes (Basel)</i> . 2022 May 3; 13(5):816. doi:10.3390/genes13050816. PMID:35627201. Bala K, Cuellar A, Herren AW, Boyadjiev SA. Identification of differentially expressed proteins between fused and open sutures in sagittal nonsyndromic craniosynostosis during suture development by quantitative proteomic analysis. <i>Proteomics Clin Appl</i> . 2021 May;15(2-3): e2000031. doi:10.1002/prca.202000031. Epub 2021 Apr 16. PMID:33580899. Calpena E, Cuellar A, Bala K, Swagemakers SMA, Koelling N, McGowan SJ, Phipps JM, Balasubramanian M, Douzgou S, Morton JEV, Shears D, Weber A, Wilson LC, Lord H, Lester T, Johnson D, Wall SA, Twigg SRF, Mathijssen IMJ, Cunningham ML, Genomics England Research Consortium, Boyadjiev SA, Wilkie AOM. SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. <i>Genet Med</i> . 2020 Jun 5;22(9):1498-1506. doi:10.1038/s41436-020-0817-2. Online ahead of print.



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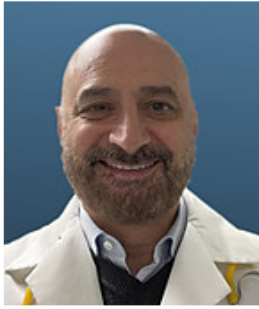
Cuellar A, Bala K, Di Pietro L, Barba M, Yagnik G, Liu JL, Stevens C, Hur DJ, Ingersoll RG, Justice CM, Drissi H, Kim J, Lattanzi W, Boyadjiev SA. Gain-of-function variants and overexpression of RUNX2 in patients with nonsyndromic midline craniosynostosis. *Bone*. 2020 Aug;137:115395. doi:10.1016/j.bone.2020.115395. Epub 2020 Apr 30. PMID:32360898.

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Justice CM, Kim J, Kim SD, Kim K, Yagnik G, Cuellar A, Carrington B, Lu CL, Sood R, Boyadjiev SA, Wilson AF. A variant associated with sagittal nonsyndromic craniosynostosis alters the regulatory function of a non-coding element. *Am J Med Genet A*. 2017 Nov;173(11):2893-2897. doi:10.1002/ajmg.a.38392. Epub 2017 Oct 6. PMID:28985029.

Zhang R, Knapp M, Suzuki K, Kajioka D, Schmidt JM, Winkler J, Yilmaz Ö, Pleschka M, Cao J, Kockum CC, Barker G, Holmdahl G, Beaman G, Keene D, Woolf AS, Cervellione RM, Cheng W, Wilkins S, Gearhart JP, Sirchia F, Di Grazia M, Ebert AK, Rösch W, Ellinger J, Jenetzky E, Zwink N, Feitz WF, Marcelis C, Schumacher J, Martín-Torres F, Hibberd ML, Khor CC, Heilmann-Heimbach S, Barth S, Boyadjiev SA, Brusco A, Ludwig M, Newman W, Nordenskjöld A, Yamada G, Odermatt B, Reutter H. ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. *Sci Rep*. 2017 Feb 8;7:42170. doi:10.1038/srep42170. PMID:28176844.

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PA, Buckley MF, Druschel CM, Mills JL, Caggana M, Romitti PA, Kay DM, Senders C, Taub PJ, Klein OD, Boggan J, Zwienenberg-Lee M, Naydenov C, Kim J, Wilson AF, Boyadjiev SA. A genome-wide association study identifies susceptibility loci for nonsyndromic sagittal craniosynostosis near BMP2 and within BBS9. *Nat Genet.* 2012 Dec;44(12):1360-4. doi:10.1038/ng.2463. Epub 2012 Nov 18. PMID:23160099.

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Boyadjiev SA, Justice CM, Eyaid W, McKusick VA, Lachman RS, Chowdry AB, Jabak M, Zwaan J, Wilson AF, Jabs EW. A novel dysmorphic syndrome with open calvarial sutures and sutural cataracts maps to chromosome 14q13-q21. *Hum Genet.* 2003 Jul;113(1):1-9. doi:10.1007/s00439-003-0932-6. Epub 2003 Apr 3. PMID:12677423.

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